AMENDMENTS TO THE CLAIMS

This listing of claims will replace all prior versions, and listings, of claims in the application. What is claimed is:

1. (currently amended) ~~A~~ An isolated nucleic acid molecule at least 20 nucleotides in length, ~~encoding a MCOLN1 polypeptide, wherein a mutation of a MCOLON1 gene encoding the MCOLN1 polypeptide results in a defect in expression of a functional MCOLN1,~~ wherein the nucleic acid shares at least ~~about~~ 95% sequence identity with a corresponding sequence from SEQ ID NO: 1 or SEQ ID NO: 2.

2. (currently amended) The nucleic acid of claim 1, ~~wherein the~~ comprising a mutation ~~is~~ selected from the group consisting of an insertion in the gene, a deletion of the gene, a truncation of the gene, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

3. (original) The nucleic acid of claim 1, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 (SEQ ID NO: 1); (b) a deletion from nucleotide 511 to nucleotide 6944 (SEQ ID NO: 1); (c) an insertion of T between nucleotide numbers 1334 and 1335 (SEQ ID NO: 2); (d) a deletion of CTT 1346-1348 (SEQ ID NO: 2); (e) an A to G substitution a position 9107 (SEQ ID NO: 1); (f) a G to T substitution at position 1461 (SEQ ID NO: 2); (g) a C to T substitution at position 429 (SEQ ID NO: 2); (h) a G to T

substitution at position 1209 (SEQ ID NO: 2); (i) a CC deletion at 598-599 (SEQ ID NO: 2); and (j) a C to T substitution at position 639 (SEQ ID NO: 2).

4. (original) The nucleic acid of claim 1, wherein the defect in expression of a functional MCOLN1 results in development of mucolipidosis IV.

5. (currently amended) The nucleic acid of claim 1, which encodes a MCOLN1 polypeptide having an amino acid sequence at least ~~about~~ 95% identical to SEQ ID NO: 3.

6. (original) The nucleic acid of claim 5, wherein the polypeptide has an amino acid sequence as depicted in SEQ ID NO: 3.

7. (original) The nucleic acid of claim 6 which has a nucleotide sequence as depicted in SEQ ID NO:1 or SEQ ID NO: 2.

Claims 8-11 are cancelled.

12. (withdrawn, currently amended) A method for detecting a genetic mutation associated with a mucolipidosis in a mammal, which method comprises using an oligonucleotide of claim 39 to detect~~ing~~ a mutation in a gene for MCOLN1, wherein the gene for MCOLN1 has a sequence at least 95% identical to SEQ ID NO: 1.

13. (withdrawn) The method according to claim 12, wherein the mutation is selected from the group consisting of an insertion in the gene, a deletion of the gene, a truncation of the gene, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

14. (withdrawn) The method according to claim 13, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 (SEQ ID NO: 1); (b) a deletion from nucleotide 511 to nucleotide 6944 (SEQ ID NO: 1); (c) an insertion of T between nucleotide numbers 1334 and 1335 (SEQ ID NO: 2); (d) a deletion of CTT 1346-1348 (SEQ ID NO: 2); (e) an A to G substitution a position 9107 (SEQ ID NO: 1); (f) a G to T substitution at position 1461 (SEQ ID NO: 2); (g) a C to T substitution at position 429 (SEQ ID NO: 2); (h) a G to T substitution at position 1209 (SEQ ID NO: 2); (i) a CC deletion at 598-599 (SEQ ID NO: 2); and (j) a C to T substitution at position 639 (SEQ ID NO: 2).

15. (withdrawn) The method according to claim 12, wherein the mucolipidosis is mucolipidosis IV.

16. (withdrawn, currently amended) A method for diagnosing a mucolipidosis, which method comprises using an oligonucleotide of claim 39 to detect~~ing~~ a mutation in a gene for MCOLN1 that results in a defect in expression of a functional MCOLN1, wherein the gene for MCOLN1 has a sequence at least 95% identical to SEQ ID NO:1.

17. (withdrawn) The method according to claim 16, wherein the mutation is selected from the group consisting of an insertion in the gene, a deletion of the gene, a truncation of the gene, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

18. (withdrawn) The method according to claim 17, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 (SEQ ID NO: 1); (b) a deletion from nucleotide 511 to nucleotide 6944 (SEQ ID NO: 1); (c) an insertion of T between nucleotide numbers 1334 and 1335 (SEQ ID NO: 2); (d) a deletion of CTT 1346-1348 (SEQ ID NO: 2); (e) an A to G substitution a position 9107 (SEQ ID NO: 1); (f) a G to T substitution at position 1461 (SEQ ID NO: 2); (g) a C to T substitution at position 429 (SEQ ID NO: 2); (h) a G to T substitution at position 1209 (SEQ ID NO: 2); (i) a CC deletion at 598-599 (SEQ ID NO: 2); and (j) a C to T substitution at position 639 (SEQ ID NO: 2).

19. (withdrawn) The method according to claim 16, wherein the mucolipidosis is MLIV.

20. (withdrawn, currently amended) A method for predicting the likelihood of developing MLIV comprising using an oligonucleotide of claim 39 to detect~~ing~~ a mutation in a gene for MCOLN1 that results in a defect in expression of a functional MCOLN1, and determining that there is a likelihood of developing MLIV if the mutation is present, wherein the gene for MCOLN4 has a sequence at least 95% identical to SEQ ID NO: 1.

21. (withdrawn) The method according to claim 20, wherein the mutation is selected from the group consisting of an insertion in the gene, a deletion of the gene, a truncation of the gene, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

22. (withdrawn) The method according to claim 21, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 (SEQ ID NO: 1); (b) a deletion from nucleotide 511 to nucleotide 6944 (SEQ ID NO: 1); (c) an insertion of T between nucleotide numbers 1334 and 1335 (SEQ ID NO: 2); (d) a deletion of CTT 1346-1348 (SEQ ID NO: 2); (e) an A to G substitution a position 9107 (SEQ ID NO: 1); (f) a G to T substitution at position 1461 (SEQ ID NO: 2); (g) a C to T substitution at position 429 (SEQ ID NO: 2); (h) a G to T substitution at position 1209 (SEQ ID NO: 2); (i) a CC deletion at 598-599 (SEQ ID NO: 2); and (j) a C to T substitution at position 639 (SEQ ID NO: 2).

23. (withdrawn, currently amended) A kit for detecting a genetic mutation in a gene for MCOLN1 that results in a defect in expression of a functional MCOLN1, comprising an oligonucleotide of claim 39 that specifically hybridizes to or adjacent to a site of a mutation of the gene for MCOLN1 that results in a defect in expression of a functional MCOLN1, wherein the gene for MCOLN1 has a sequence at least 95% identical to SEQ ID NO: 1.

24. (withdrawn) The kit according to claim 23, wherein the oligonucleotide is a labeled probe having a sequence corresponding to the sequence of the gene encoding MCOLN1 at the site of the mutation, whereby hybridization of the probe is indicative of the presence of the mutation.

25. (withdrawn) The kit according to claim 23, wherein the oligonucleotide hybridizes to a first site adjacent to the site of the mutation, further comprising a second oligonucleotide that specifically hybridizes to a second site adjacent to the site of the mutation, wherein the second site is on the opposite strand relative to the first site, and oriented relative to the first site such that both sites flank opposite sides of the site of the mutation, whereby the first and second oligonucleotides serve as primers for PCR amplification of the site of the mutation.

26. (withdrawn) The kit according to claim 23, wherein the mutation is selected from the group consisting of an insertion in the gene, a deletion of the gene, a truncation of the gene, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

27. (withdrawn) The kit according to claim 26, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 (SEQ ID NO: 1); (b) a deletion from nucleotide 511 to nucleotide 6944 (SEQ ID NO: 1); (c) an insertion of T between nucleotide numbers 1334 and 1335 (SEQ ID NO: 2); (d) a deletion of CTT 1346-1348 (SEQ ID NO: 2); (e) an A to G substitution a position 9107 (SEQ ID NO: 1); (f) a G to T substitution at position 1461 (SEQ ID NO: 2); (g) a C to T substitution at position 429 (SEQ ID NO: 2); (h) a G to T

substitution at position 1209 (SEQ ID NO: 2); (i) a CC deletion at 598-599 (SEQ ID NO: 2); and (j) a C to T substitution at position 639 (SEQ ID NO: 2).

Claims 28-32 are cancelled.

33. (withdrawn, currently amended) An expression vector comprising a gene encoding functional human MCOLN1 operatively associated with a promoter, wherein the functional MCOLN1 has an amino acid sequence that is at least ~~about~~ 95% identical to SEQ ID NO: 3.

34. (withdrawn) The expression vector of claim 33, wherein the functional MCOLN1 has an amino acid sequence as depicted in SEQ ID NO: 3.

35. (withdrawn) A pharmaceutical composition comprising the expression vector of claim 33 and a pharmaceutically acceptable carrier or excipient.

Claims 36-38 are cancelled.

39. (new) The nucleic acid of claim 1, wherein the nucleic acid is a single stranded oligonucleotide.